



<110>	Hageman	ı,	Grego	s.	
	Kuehn,	Ma	arkus	Η.	

<120> THERAPEUTICS AND DIAGNOSTICS FOR OCULAR DISEASE BASED ON A NOVEL HUMAN GENE FAMILY

<130> UIA-027.01

<140> 09/183,972

<141> 1998-10-29

<160> 49

<170> PatentIn Ver. 2.0

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Cys Leu Phe Asp Ile Gly Gln Asn Phe Ser Asn Ser Gln Glu His Leu

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Asp Leu Leu Gln Gln Arg Ile Lys Gln Arg Ser Phe Pro Glu Arg Lys

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Asp Glu Val Ser Thr Glu Lys Thr Leu Gly Glu Pro Ser Glu Thr Ile
100 105 110

gtg gtt tca aca gat gtt gcc agc gtc tca ctt ggg cct ttc cct gtc 384 Val Val Ser Thr Asp Val Ala Ser Val Ser Leu Gly Pro Phe Pro Val 115 120 125

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Thr	Gly 50	Glu	Tyr	Gln	Asp	Trp 55	Val	Ser	Phe	Cys	Gln 60	Gln	Glu	Thr	Phe	
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Thr	Pro 130	Asp	Asp	Thr	Leu	Leu 135	Asn	Glu	Ile	Leu	Asp 140	Asn	Ala	Leu	Asn	
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	Tyr					Glu		gac Asp			Pro					1177
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Thr Lys Asp Ile Asp Asn Pro Pro Arg Asn Glu Thr Thr Glu Ser Thr 35 40 45

Glu Lys Met Tyr Lys Met Ser Thr Met Arg Arg Ile Phe Asp Leu Ala
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Lys His Arg Thr Lys Arg Ser Ala Phe Phe Pro Thr Gly Val Lys Val
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Tyr Arg Leu Arg Val Cys Gln Glu Ala Val Trp Glu Ala Tyr Arg Ile
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Arg Ser Phe Pro Asp Arg Lys Asp Glu Ile Ser Ala Glu Lys Thr Leu 165 170 175

Gly Glu Pro Gly Glu Thr Ile Val Ile Ser Thr Asp Val Ala Asn Val
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Gln Thr Tyr Leu Ser Ile Glu Glu ‡le Gln Glu Pro Lys Ser Ala Val
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Ser Phe Leu Leu Pro Glu Glu Ser/ Thr Asp Leu Ser Leu Ala Thr Lys
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aag aaa cag cct ctg gac cgc aga gaa act gaa aga cag tgg tta atc
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Lys Lys Gln Pro Leu Asp Arg Arg Glu Thr Glu Arg Gln Trp Leu Ile
aga agg cgg aga tot att otg tit oot aat gga gtg aaa ato tgo oca
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Arg Arg Arg Ser Ile Leu Phe Pro Asn Gly Val Lys Ile Cys Pro
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Asp Glu Ser Val Ala Glu Ala Val Ala Asn His Val Lys Tyr Phe Lys
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Val Arg Val Cys Gln Glu Ala Val Trp Glu Ala Phe Arg Thr Phe Trp
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Glu Asp Gly Val Thr Ser Ile the Glu Met Gly Thr Asn Phe Ser Glu
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					tct Ser											1334

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ccc Pro					Ġly				234
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aag Lys									253
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tct Ser 815									267

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			atc ctc aac (/Ile Leu Asn)		
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		15		
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atg cct ggg cac ggg g Met Pro Gly His Gly A 1070 10				46
tgg tac cga ggc aag c Trp Tyr Arg Gly Lys H 1090	lis Cys Glu Glu			94
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tcctgttctg aaactgatta	ı gaagcetgga gaa	agatggag attacttgt	t acttatgtca 398	84
tataattaac ctggatttta	aacactgttg gaa	agaagagn tttctatga	a aaaattaaat 404	44
atagggcaca ctgtttttt	ttcagettaa gn	ttcagaa tgtagtnag	a gatgtwmcat 410	04
ttttatttct ataaagactg	g aatgetgtgt tta	aaataatt gaaaactac	g ttaaaaaaaa 410	64
a			410	65

<210> 6 <211> 1241 <212> PRT <213> Homo sapiens <400> 6 Met Ile Met Phe Pro Leu Phe Gly Lys Ile Ser Leu Gly Ile Leu Ile Phe Val Leu Ile Glu Gly Asp Phe Pro Ser Leu Thr Ala Gln Thr Tyr Leu Ser Ile Glu Glu Ile Gln Glu Pro Lys Ser Ala Val Ser Phe Leu 40 Leu Pro Glu Glu Ser Thr Asp Leu Ser Leu Ala Thr Lys Lys Gln 55 Pro Leu Asp Arg Arg Glu Thr Gl/u Arg Gln Trp Leu Ile Arg Arg Arg 70 Arg Ser Ile Leu Phe Pro Asn Gly Val Lys Ile Cys Pro Asp Glu Ser Val Ala Glu Ala Val Ala Asn His Val ∕Lys Tyr Phe Lys Val Arg Val 102 Cys Gln Glu Ala Val Trp Glu Ala Phe Arg Thr Phe Trp Asp Arg Leu 120 Pro Gly Arg Glu Glu Tyr His Tyr Trp Met Asn Leu Cys Glu Asp Gly Val Thr Ser Ile Phe Glu Met Gly Thr Asn Phe Ser Glu Ser Val Glu His Arg Ser Leu Ile Met Lys Lys Leu Thr Tyr Ala Lys Glu Thr Val Ser Ser Ser Glu Leu Ser Ser Pro Val Pro Val Gly Asp Thr Ser Thr 185 Leu Gly Asp Thr Thr Leu Ser Val Pro His Pro Glu Val Asp Ala Tyr 195 200 Glu Gly Ala Ser Glu Ser Ser Leu Glu Arg Pro Glu Glu Ser Ile Ser 215 Asn Glu Ile Glu Asn Val Ile Glu Glu Ala Thr Lys Pro Ala Gly Glu 225 230 235 Gln Ile Ala Glu Phe Ser Ile His Leu Leu Gly Lys Gln Tyr Arg Glu Glu Leu Gln Asp Ser Ser Ser Phe His His Gln His Leu Glu Glu Glu

265

Phe Ile Ser Glu Val Glu Asn Ala Phe Thr Gly Leu Pro Gly Tyr Lys 280 275 Glu Ile Arg Val Leu Glu Phe Arg Ser Pro Lys Glu Asn Asp Ser Gly 295 Val Asp Val Tyr Tyr Ala Val Thr Phe Asn Gly Glu Ala Ile Ser Asn Thr Thr Trp Asp Leu Ile Ser Leu His Ser! Asn Lys Val Glu Asn His 325 330 Gly Leu Val Glu Leu Asp Asp Lys Pro Thr Val Val Tyr Thr Ile Ser 345 Asn Phe Arg Asp Tyr Ile Ala Glu Thr Leu Gln Gln Asn Phe Leu Leu 360 365 Gly Asn Ser Ser Leu Asn Pro Asp Pro Asp Ser Leu Gln Leu Ile Asn 375 **B80** Val Arg Gly Val Leu Arg His Gln Thr Glu Asp Leu Val Trp Asn Thr 395 Gln Ser Ser Ser Leu Gln Ala Thr Pro Ser Ser Lle Leu Asp Asn Thr 410 Phe Gln Ala Ala Trp Pro Ser Ala Asp Glu Ser Ile Thr Ser Ser Ile 425 Pro Pro Leu Asp Phe Ser Ser Gly Pto Pro Ser Ala Thr Gly Arg Glu Leu Trp Ser Glu Ser Pro Leu Gly Asp/ Leu Val Ser Thr His Lys Leu 450 Ala Phe Pro Ser Lys Met Gly Leu Ser Ser Ser Pro Glu Val Leu Glu 475 Val Ser Ser Leu Thr Leu His Ser Val Thr Pro Ala Val Leu Gln Thr 485 490 Gly Leu Pro Val Ala Ser Glu Glu Arg Thr Ser Gly Ser His Leu Val Glu Asp Gly Leu Ala Asn Val Glu Glu Ser Glu Asp Phe Leu Ser Ile 515 520 Asp Ser Leu Pro Ser Ser Ser Phe Thr Gln Pro Val Pro Lys Glu Thr Ile Pro Ser Met Glu Asp Ser Asp Val Ser Leu Thr Ser Ser Pro Tyr 555 Leu Thr Ser Ser Ile Pro Phe Gly Leu Asp Ser Leu Thr Ser Lys Val 565 570 575

Lys Asp Gln Leu Lys Val Ser Pro Phe Leu Pro Asp Ala Ser Met Glu 585 Lys Glu Leu Ile Phe Asp Gly Gly Leu Gly Ser Gly Ser Gly Gln Lys Val Asp Leu Ile Thr Trp Pro Trp Ser Glu Thr Ser Ser Glu Lys Ser 615 Ala Glu Pro Leu Ser Lys Pro Trp Leu Glu Asp Asp Ser Leu Leu 630 Pro Ala Glu Ile Glu Asp Lys Lys Leu Val Leu Val Asp Lys Met Asp 650 Ser Thr Asp Gln Ile Ser Lys His \$er Lys, Tyr Glu His Asp Asp Arg 665 Ser Thr His Phe Pro Glu Glu Glu Pro Leu Ser Gly Pro Ala Val Pro 680 Ile Phe Ala Asp Thr Ala Ala Glu |Se/r Ala Ser Leu Thr Leu Pro Lys 695 His Ile Ser Glu Val Pro Gly Val Asp Asp Cys Ser Val Thr Lys Ala 710 715 Pro Leu Ile Leu Thr Ser Val Ala tle Ser Ala Ser Thr Asp Lys Ser 730 Asp Gln Ala Asp Ala Ile Leu Arg Glu Asp Met Glu Gln Ile Thr Glu 745 Ser Ser Asn Tyr Glu Trp Phe Asp Ser Glu Val Ser Met Val Lys Pro Asp Met Gln Thr Leu Trp Thr Ile Leu Pro Glu Ser Glu Arg Val Trp Thr Arg Thr Ser Ser Leu Glu Lys Leu Ser Arg Asp Ile Leu Ala Ser 790 795 800 Thr Pro Gln Ser Ala Asp Arg Leu Trp Leu Ser Val Thr Gln Ser Thr 810 Lys Leu Pro Pro Thr Thr Ile Ser Thr Leu Leu Glu Asp Glu Val Ile 820 825 Met Gly Val Gln Asp Ile Ser Leu Glu Leu Asp Arg Ile Gly Thr Asp Tyr Tyr Gln Pro Glu Gln Val Gln \$lu Gln Asn Gly Lys Val Gly Ser 855 Tyr Val Glu Met Ser Thr Ser Val His Ser Thr Glu Met Val Ser Val 870 865 875

Ala Trp Pro Thr Glu Gly Gly Asp Asp Leu Ser Tyr Thr Gln Thr Ser 890 Gly Ala Leu Val Val Phe Phe Ser Leu Arg Val Thr Asn Met Met Phe Ser Glu Asp Leu Phe Asn Lys Asn Ser Leu Glu Tyr Lys Ala Leu Glu Gln Arg Phe Leu Glu Leu Leu Val Pro Tyr Leu Gln Ser Asn Leu Thr 935 Gly Phe Gln Asn Leu Glu Ile Leu Asn Phe Arg Asn Gly Ser Ile Val 950 955 Val Asn Ser Arg Met Lys Phe Ala Asn Ser Val Pro Pro Asn Val Asn 970 965 Asn Ala Val Tyr Met Ile Leu Glu Asp Phe Cys Thr Thr Ala Tyr Asn 98/5 Tyr Ser Leu Asp Val Glu Ser Gly Thr Met Asn Leu Ala Ile Asp Lys 1000 Asp Glu Ala Asn Pro Cys Lys Phe Gln Ala Cys Asn Glu Phe Ser Glu 1015 1020 Cys Leu Val Asn Pro Trp Ser Gly \$\text{flu Al}\a Lys Cys Arg Cys Phe Pro 1030 1035 Gly Tyr Leu Ser Val Glu Glu Arg Pro Cys Gln Ser Leu Cys Asp Leu 1045 1050 Gln Pro Asp Phe Cys Leu Asn Asp Gly Lys Cys Asp Ile Met Pro Gly 1060 His Gly Ala Ile Cys Arg Cys Arg Val Gly Glu Asn Trp Trp Tyr Arg Gly Lys His Cys Glu Glu Phe Val Ser Glu Pro Val Ile Ile Gly Ile 1090 1095 Thr Ile Ala Ser Val Val Gly Leu Leu Val Ile Phe Ser Ala Ile Ile 1110 1115 Tyr Phe Phe Ile Arg Thr Leu Gln Ala His His Asp Arg Ser Glu Arg 1125 1130 1135 Glu Ser Pro Phe Ser Gly Ser Ser Arg Gln Pro Asp Ser Leu Ser Ser 1145 Ile Glu Asn Ala Val Lys Tyr Asn Pro Val Tyr Glu Ser His Arg Ala 1160 Gly Cys Glu Lys Tyr Glu Gly Pro Tyr Pro Gln His Pro Phe Tyr Ser 1170 1175

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Ser Ala Ser Gly Asp Val Ile Gly Gly Leu Ser Arg Glu Glu Ile Arg
                                       1195
185
                   1190
Gln Met Tyr Glu Ser Ser Glu Leu Ser Arg Glu Glu Ile Gln Glu Arg
               1205
                                    1210
Met Arg Val Leu Glu Leu Tyr Ala Asn Asp Pro Glu Phe Ala Ala Phe
                                1225
Val Arg Glu Gln Gln Val Glu Glu Val
                           1240
<210> 7
<211> 20
<212> PRT
<213> rattus
<400> 7
Ser Ile Leu Phe Pro Asn Gly Val Arg Ile Cys Pro Ser Asp Thr Val
                                      10
Ala Glu Ala Val
<210> 8
<211> 20
<212> PRT
<213> porcine
<220> ·
<221> UNSURE
<222> (1)..(20)
<223> applicants are unsure of residues designated as
      "Xaa" at positions 1 & 11
Xaa Val Phe Phe Pro Thr Gly Val Lys Val Xaa Pro Gln Glu Ser Met
                                      10
Lys Gln Ile Leu
             20
<210> 9
<211> 10
<212> PRT
<213> porcine
<220>
<221> UNSURE
<222> (1)..(10)
<223> applicants are unsure of residues designated as
      "Xaa" at position 1
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<400> 9
Xaa Val Leu Phe Pro Asn Gly Val Lys/Ile
<210> 10
<211> 30
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<220>
<221> modified base
<222> (25)
<223> i
<400> 10
                                                                    30
tattaggaat tccatyttyt tyccnaaygg
<210> 11
<211> 26
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<220>
<221> modified_base
<222> (1)..(26)
<223> "n" at positions 3, 6, 9 & 24 represent "inosine"
<400> 11
                                                                    26
ttnccngcna gytcytgrta rtangg
<210> 12
<211> 22
<212> DNA
<213> Artificial Sequence
<220>
<223> Description of Artificial Sequence: primer
<400> 12
                                                                    22
ggatttttct ccaagttcaa gg
<210> 13
<211> 20
<212> DNA
<213> Artificial Sequence
<220>
<223> Description of Artificial Sequence: primer
```

```
<400> 13
                                                                    20
acggggtta aagtctgtcc
<210> 14
<211> 20
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<400> 14
                                                                    20
cgaacaaaa gatccgcatt
<210> 15
<211> 21
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence,
                                          'primer
<400> 15
ccttctgcct ctttgacatt g
                                                                    21
<210> 16
<211> 20
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<400> 16
atcaggactg ggtcagcatc
                                                                    20
<210> 17
<211> 20
<212> DNA
<213> Artificial Sequence
<220>
<223> Description of Artificial Sequence: primer
<400> 17
                                                                    20
tcaacagatg ttgccaacgt
<210> 18
<211> 20
<212> DNA
<213> Artificial Sequence
```

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<220>
<223> Description of Artificial Sequence: primer
<400> 18
                                                                    20
gagcctggtg aaaccattgt
<210> 19
<211> 20
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial $equence: primer
<400> 19
                                                                    20
gtggagctca gcgtctctct
<210> 20
<211> 19
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial $equence:
                                           primer
<400> 20
                                                                    19
tgtgttggag gagcagagg
<210> 21
<211> 20
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<400> 21
aaaagatggc tcaagctcca
                                                                    20
<210> 22
<211> 22
<212> DNA
<213> Artificial Sequence
<220>
<223> Description of Artificial Sequence: primer
<400> 22
                                                                    22
gaaacttcca ggattcaaaa aa
<210> 23
<211> 20
<212> DNA
```

√ 24`		,
<213> Artificial Sequence		
<220>		
<223> Description of Artificial Sequence: primer		
<400> 23		
aggaggacaa gcaaccagaa	20	
<210> 24		
<211> 22		
<212> DNA		
<213> Artificial Sequence		
<220>		
<223> Description of Artificial Sequence: primer		
<400> 24		
tccaacaaaa ttgaaagtga gg	22	
/		
<210> 25		
<211> 20		
<211> 20 <212> DNA		
<213> Artificial Sequence		
(213) Altilitial Sequence		
<220>		
<223> Description of Artificial Sequence: primer		
1400- 25		
<400> 25 aatcagagct gcccacatct	20	
<210> 26		
<210> 26 <211> 19		
<211> 19 <212> DNA		
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<220> <223> Description of Artificial Sequence: primer		
<400> 26	3.0	
agcctttggt cctgacacc	19	
<210> 27 <211> 21		
<211> 21 <212> DNA		
<212> DNA <213> Artificial Sequence		
22137 Metiticial bequence		
<220>		
<223> Description of Artificial Sequence: primer		
<400> 27		
ccacctttct ttatggcatc a	21	
<210> 28		

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<211> 20 ·
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<400> 28
agtgcaggtg gcgaagatat
                                                                    20
<210> 29
<211> 20
<212> DNA
<213> Artificial Sequence
<223> Description of Artificia Sequence: primer
<400> 29
ctccctgtca gaagctccac
                                                                    20
<210> 30
<211> 22
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<400> 30
ccacctgcat cttcagatga ca
                                                                    22
<210> 31
<211> 20
<212> DNA
<213> Artificial Sequence
<220>
<223> Description of Artificial Sequence: primer
<400> 31
agttctatga ccattgcccc
                                                                    20
<210> 32
<211> 22
<212> DNA
<213> Artificial Sequence
<220>
<223> Description of Artificial Sequence: primer
<400> 32
aggataccac tcctgtctca gc
                                                                    22
```

```
<210> 33
<211> 21
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<400> 33
aacaattcac acagctgctg g
                                                                    21
<210> 34
<211> 17
<212> DNA
<213> Artificial Sequence
<223> Description of Artificia | Sequence: primer
<400> 34
ccgagctctg gagcaac
                                                                    17
<210> 35
<211> 20
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<400> 35
                                                                    20
gaggattttc gttctgctgc
<210> 36
<211> 20
<212> DNA
<213> Artificial Sequence
<220>
<223> Description of Artificial Sequence: primer
<400> 36
                                                                    20
tctgtgccgt ataacctcac
<210> 37
<211> 20
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<400> 37
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gactgaggaa gcggagtgtc	20
<210> 38 <211> 18 <212> DNA <213> Artificial Sequence	
<220> <223> Description of Artificial Sequence: primer	
<400> 38 acgaacggac tgaggaag	18
<210> 39 <211> 22 <212> DNA <213> Artificial Sequence	
<220> <223> Description of Artificial Sequence: primer	
<400> 39 ttctgaatta ctgaccgtag aa	22
<210> 40 <211> 22 <212> DNA <213> Artificial Sequence	
<220> <223> Description of Artificial Sequence: primer	
<400> 40 ttccaaaatc aacaaataa ca	22
<210> 41 <211> 21 <212> DNA <213> Artificial Sequence	
<220> <223> Description of Artificial Sequence: primer	
<400> 41 ggtcatcaaa atccagacat a	21
<210> 42 <211> 27 <212> DNA <213> Artificial Sequence	
<220> <223> Description of Artificial Sequence: primer	

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<400> 42
tgccttctca aggaaaatgg agacagg
                                                                    27
<210> 43
<211> 20
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial Sequence: primer
<400> 43
                                                                    20
taagccaggt ttgcttccac
<210> 44
<211> 20
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial $equence: primer
.<400> 44
taaaacccca aatgcaatca
                                                                    20
<210> 45
<211> 21
<212> DNA
<213> Artificial Sequence
<223> Description of Artificial $equence: primer
<400> 45
gcaggtctct ctaaacgcat g
                                                                    21
<210> 46
<211> 15
<212> PRT
<213> Homo sapiens
<220>
<221> UNSURE
<222> (1)..(15)
<223> applicants are unsure of residues designated as
      "Xaa" at positions 1 & 11
<400> 46
Xaa Ala Leu Phe Pro Asn Gly Val Leu Ile Xaa Pro Xaa Glu Val
  1
                  5
                                      10
                                                           15
<210> 47
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<211> 19
<212> PRT
<213> Callimico sp.
<220>
<221> UNSURE
<222> (1)..(19)
<223> applicants are unsure of residues designated as
      "Xaa" at positions 1, 11 & 16
Xaa Ile Leu Phe Pro Asn Gly Val Leu Ile Xaa Pro Asp Glu Val Xaa
                                      10
Lys Glu Ile
<210> 48
<211> 20
<212> PRT
<213> Homo sapiens
<400> 48
Ser Ala Phe Phe Pro Thr Gly Val Lys Val Cys Pro Gln Glu Ser Met
                                     10
                  5
Lys Gln Ile Leu
             20
<210> 49
<211> 18
<212> PRT
<213> Callimico sp.
<220>
<221> UNSURE
<222> (1)..(18)
<223> applicants are unsure of residues designated as
      "Xaa" at positions 1 & 11
<400> 49
Xaa Ile Phe Pro Thr Gly Val Lys Val Xaa Pro Gln Glu Ser Met
  1
Lys Gln
```

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